

Getting to the Point: What Women Newly Diagnosed With Breast Cancer Want to Know About Treatment-Focused Genetic Testing

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Traditionally, genetic testing for a germ-line mutation in the breast cancer protection genes (i.e., *BRCA1* or *BRCA2*) in women with a breast cancer diagnosis has been confined to those with a family history of breast and/or ovarian cancer. Testing of such women usually takes place after completion of active cancer treatment with the purpose of guiding their future risk management in relation to a new primary breast and/or ovarian cancer diagnosis. In addition, genetic risk information has significant risk management implications for a woman's unaffected genetic relatives who may themselves consider predictive genetic testing to clarify their risk of breast and/or ovarian cancer.

However, growing evidence shows that mutation status may influence breast cancer management recommendations and that benefits may arise from having genetic counseling and testing available shortly after a cancer diagnosis (Silva, 2008; Trainer et al., 2010; Tutt & Ashworth, 2008). The process, referred to in this article as treatment-focused genetic testing (TFGT), may help guide a woman's initial cancer treatment and future risk management. In the first instance, TFGT may assist with the complex decision-making process regarding a woman's surgical options, including the selection of breast conservation or a therapeutic mastectomy, with or without a contralateral risk-reducing mastectomy, based on the patient's mutation status and/or family history of cancer (Meiser et al., 2008).

In addition, evidence suggests that *BRCA*-associated tumors are more sensitive to platinum agents and potentially less resistant to taxanes (Quinn et al., 2007; Rottenberg et al., 2007; Trainer et al., 2010). Novel agents also are being tested that target *BRCA* tumors;

Purpose/Objectives: To identify young women's information preferences regarding treatment-focused genetic testing (TFGT) and to develop and evaluate a novel educational resource.

Research Approach: Qualitative interview study and pilot testing of a novel resource.

Setting: Two familial cancer services and one outpatient oncology clinic in Sydney and Melbourne, Australia.

Participants: 26 women with breast cancer aged 50 years and younger who either previously had TFGT ($n = 14$) or had a diagnosis of breast cancer within the previous 6–12 months.

Methodologic Approach: Participants were asked about their views of TFGT in semistructured interviews. A brief pamphlet on TFGT then was developed and pilot tested with 17 of the 26 women.

Main Research Variables: Women's attitudes and preferences with regard to timing, mode of delivery, and amount and format of information regarding TFGT were explored.

Findings: Most women wanted to be informed about TFGT at or around the time of their cancer diagnosis via a face-to-face consultation. No clear preference existed for which type of healthcare professional should provide information on TFGT. Brief written information about TFGT was viewed as important supporting material. The educational resource developed was well received.

Conclusions: The potential for more widespread TFGT in the future indicates a need for patient educational materials that enable women to make informed choices about TFGT. This pilot study has provided timely initial evidence on the efficacy of a brief written resource in preparing women for decision making about TFGT.

Interpretation: The resource developed in this study will assist oncology nurses to make important genetic risk information available to women newly diagnosed with breast cancer at a stressful time.

specifically poly (ADP-ribose) polymerase inhibitors that have been found to be **active agents with high response rates** in patients with recurrent disease (Fong et al., 2009, 2010). Therefore, as more conclusive evidence on the efficacy of conventional chemotherapy and novel agents becomes available, TFGT is likely to be increasingly used to tailor women's cancer treatments.

Rapid advances in sequencing technology are likely to decrease the cost of genetic testing and the timeframe within which results can be provided (Klee, Hoppman-Chaney, & Ferber, 2011). In addition, the traditional approach of using family history as the major selection criteria for genetic testing is being challenged, as the proportion of *BRCA1* and *BRCA2* mutation carriers found in patients with early onset breast cancer without a strong family history has ranged from 6% (Lalloo et al., 2003) to 78% (Choi, Lee, Bale, Carter, & Haffty, 2004). **As such, a pressing need exists for data on effective educational strategies regarding TFGT for women newly diagnosed with breast cancer, both with and without a strong family history, in advance of new technology being widely implemented into clinical practice.**

Background

Very little currently is known about the acceptability of TFGT among women with and without a strong family history of breast and/or ovarian cancer and their associated information needs. Two prospective studies conducted in the United States and the Netherlands have assessed the psychological and behavioral impact of TFGT (Schlich-Bakker et al., 2006; Schwartz et al., 2004). The Dutch study assessed the psychological impact of TFGT in women with breast cancer who were shortly to commence adjuvant radiotherapy. The authors found that distress levels did not increase after the offer of genetic counseling and testing (Schlich-Bakker et al., 2006). A small study in the United Kingdom, using a focus group methodology, explored whether women diagnosed with breast cancer before the age of 40 wanted information about genetic testing close to the time of diagnosis. All 13 participants already had been identified as *BRCA* carriers (Ardern-Jones, Kenen, & Eeles, 2005). The majority of women felt that an offer of genetic testing around the time of their cancer diagnosis would have been too stressful, although some women reported that the offer would be important if it had the potential to alter treatment decisions. All women agreed that there was no one right time for everyone. An important limitation of those studies is that they almost exclusively included women with a strong family history of breast and/or ovarian cancer.

In a qualitative study, Vadaparampil et al. (2009) assessed the impact of a surgeon referral letter on patients recently diagnosed with breast cancer and their uptake

of *BRCA* genetic counseling and testing. Many women who had been sent a letter by their surgeon for *BRCA* genetic counseling reported mixed reactions to the letter, and some women were confused or concerned about why they had received a letter. In addition, about 20% did not recall receiving the letter. Based on those findings, the authors concluded that a referral letter from a surgeon may not be the most effective means of informing patients about TFGT. The authors suggested that the letter may have been more effective if it had included more detailed information on the surgical implications of *BRCA* testing and on the process of TFGT; that suggestion was, however, not based on data regarding women's preferences relating to the content and format of the educational materials.

To the authors' knowledge, no educational resources currently are available specifically for women newly diagnosed with breast cancer that provide information about TFGT to assist with informed decision making. Detailed decision aids have been developed for people affected by cancer considering genetic testing after completion of their cancer treatment, and for unaffected people at increased risk of hereditary cancer considering predictive testing (Gaff & Meiser, 2009; Green, Biesecker, McInerney, Mauger, & Fost, 2001; Green et al., 2004, 2005; Mancini et al., 2006; Schwartz et al., 2001; van Roosmalen et al., 2004; Wakefield, Meiser, Homewood, Peate, et al., 2008; Wakefield, Meiser, Homewood, Ward, et al., 2008). Decision aids have been shown to be effective in meeting the information needs of those specific populations (Gaff & Meiser, 2009). However, TFGT among young women with breast cancer is very different compared to genetic testing offered to patients following completion of their cancer treatment and to predictive testing, where knowledge of mutation status does not influence treatment. Therefore, ascertaining the specific information needs (regarding TFGT) of patients newly diagnosed with breast cancer is critical to the development of appropriate educational materials that will not overwhelm the patients at a highly stressful time. Specifically, data regarding the content, format, and mode of delivery of information about TFGT will inform the development of high-quality educational materials.

The current study was carried out in two stages. First, in-depth interviews were conducted to identify the information and communication needs regarding TFGT of women newly diagnosed with breast cancer who were age 50 or younger. The age cutoff was chosen because younger women may be more likely to use mastectomy for future prevention, as they have more life-years at risk. The views of young women with and without a strong family history of breast or ovarian cancer were included because both groups are likely to be targeted for TFGT. On the basis of the findings

from the qualitative study in stage 1, the authors then developed and pilot tested a psychoeducational resource in stage 2 to enable women newly diagnosed with breast cancer to make informed decisions about genetic testing for germ-line *BRCA* mutations and to facilitate discussions with their healthcare professionals. Because the development of the educational resource was guided by women's preferences identified in in-depth interviews about TFGT conducted in stage 1, the authors expected that the materials would be evaluated equally favorably by women who had already undergone TFGT and by those who had not. The authors predicted, therefore, no differences between the two groups of women in terms of their satisfaction with the resource, the emotional impact of reading the material, the perceived importance of TFGT, or in perceived improvement in understanding of TFGT.

Methodologic Approach

Participants

Two different groups of women with breast cancer (age 50 and younger) were recruited. Group A was comprised of women, ascertained through two family cancer clinics in Sydney and Melbourne, Australia, who already had undergone TFGT to facilitate surgical decisions (Group A denotes actual decision making about TFGT); all had a strong family history of breast and/or ovarian cancer according to national guidelines (National Breast Cancer Centre, 2006). Group H was comprised of women who were unselected for family history and diagnosed with breast cancer within the previous 6–12 months at an oncology clinic in Sydney (Group H denotes hypothetical decision making about TFGT). Exclusion criteria included having had a breast cancer recurrence to avoid undue participant burden, being younger than 18 years of age, having insufficient English language skills to complete the interview unaided, or having obvious intellectual or mental

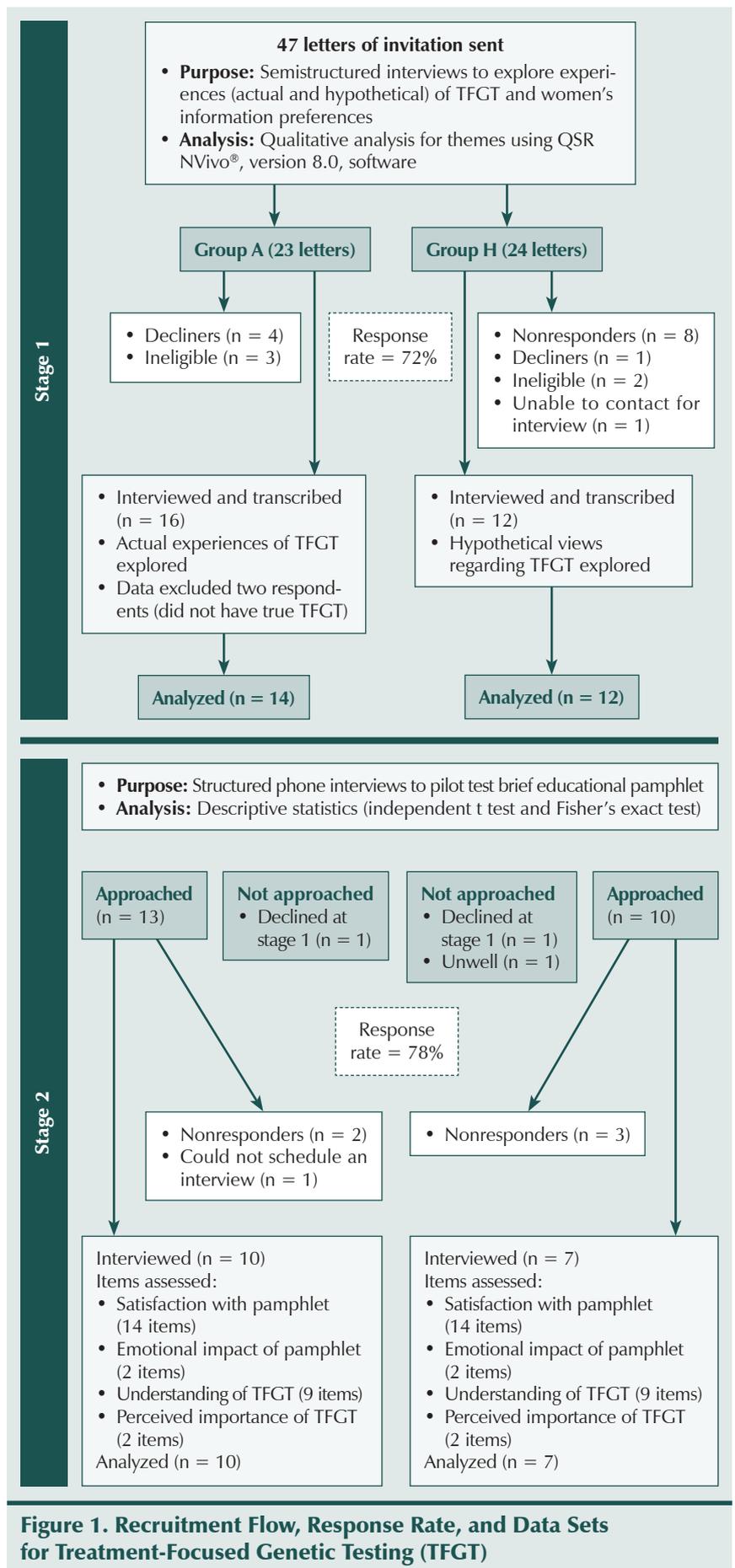


Figure 1. Recruitment Flow, Response Rate, and Data Sets for Treatment-Focused Genetic Testing (TFGT)

impairment. For both groups, a letter of invitation to participate in the study interview was sent by the treating clinician. The study process is presented in Figure 1. Forty-seven letters of invitation were mailed, with 34 women opting into the study (response rate of 72%). Of those women, five were ineligible and one woman could not be contacted for an interview. The data for 2 of the 28 women interviewed were excluded because it was ascertained during their interview that they had genetic counseling after their definitive breast cancer treatment. Approval was obtained from the relevant human research ethics committees (South Eastern Sydney Local Health District and Peter MacCallum Cancer Centre).

Stage 1: Qualitative Analyses

Data collection: Prior to the telephone interview, women were mailed a consent form, a one-page information sheet regarding TFGT, and a decision aid about another topic (as an example of one type of educational material) to elicit preferences for specific information presentation. The sample decision aid included a personal worksheet designed to elicit the perceived pros and cons of particular management options (O'Connor et al., 1998).

A qualitative data collection method was used to identify the range of preferences about information provision (Denzin & Lincoln, 1994). The interviews were semistructured with probes to elicit more information when appropriate. Questions explored women's attitudes and preferences with regard to timing and mode of delivery and format of information regarding TFGT. Results from early interviews were used to suggest additional lines of questioning in subsequent interviews to ensure that divergent points of view were explored (Miles & Huberman, 1994). All interviews were conducted by a researcher who has extensive experience both as a cancer genetic counselor and as an oncology nurse. Sampling was discontinued at the point when data saturation was reached (Denzin & Lincoln, 1994).

Data analysis: The conceptual framework of Miles and Huberman (1994) was used to guide the analysis. Two researchers identified the initial themes and categories and coded two transcripts concurrently to further refine themes and categories; if discrepancies occurred with respect to specific categories, discussions took place until consensus was achieved. A researcher then coded the transcripts using the qualitative data analysis software QSR NVivo®, version 8.0, to categorize the data and to facilitate systematic comparisons based on participant characteristics, including participant group (Group A versus Group H) and whether or not a woman had children. Another researcher conducted all data analyses, using QSR NVivo, and wrote the descriptive text on the findings. The use of multiple coders and analysts is recommended by Miles and

Huberman (1994) to reduce the potential for researcher bias and to increase the validity of the findings.

Stage 2: Development and Pilot Evaluation of the Pamphlet

Procedure: The one-page bifold pamphlet was designed to provide basic information about TFGT and to facilitate women's discussions with their healthcare professional(s) about TFGT. The pamphlet provided information about TFGT, including why women may wish to consider it, what it involves, and the potential consequences and implications of TFGT results. The early prototypes were developed iteratively involving a multidisciplinary committee, including researchers and clinicians with expertise in clinical genetics, genetic counseling, genetics education, oncology, and psychology. Readability of the pamphlet was adjusted to a ninth-grade level (National Health and Medical Research Council, 1999). Women who participated in stage 1 were invited to participate in stage 2 interviews, which evaluated the acceptability and impact of the pamphlet. Women who wished to be involved in stage 2 were interviewed by telephone by a member of the research team who was not involved in the development of the materials.

Measures

The following items were included in the stage 2 interview and were based on similar items used in previous related studies (Peate et al., 2009; Wakefield et al., 2007). Fourteen items evaluated the amount of information provided, perceived usefulness, and satisfaction with the pamphlet in a combination of structured categorical (see Table 1) and open-ended responses. Women also were asked to identify areas that required more or less detail, and to specify what they liked best and least about the pamphlet. Nine items (see Table 2) assessed the perceived extent to which the pamphlet would have improved women's understanding of TFGT. The response options ranged from 1 (not at all) to 5 (a lot). Participants' responses were summed to provide a total score (range of 9–40).

Participants were asked to rank how much the pamphlet had made them feel worried or concerned, or upset or sad using two five-point scales ranging from 1 (not at all) to 5 (very much so). Importance of TFGT was determined using two three-point rating scales ranging from 1 (not at all important) to 3 (very important). Participants were asked to indicate how important they felt TFGT would have been at the time of diagnosis and their perceptions of the importance attributed to TFGT by their clinician. Women also were asked to, "imagine that [the pamphlet] had been given to you around the time of your breast cancer

diagnosis. Would this pamphlet have been enough for you to make a decision about whether or not to have testing?"

Results

Stage 1: Qualitative Analyses

Women (N = 26) with breast cancer (age 50 or younger) who either previously had TFGT (Group A, n = 14) or had been diagnosed with breast cancer in the previous 6–12 months and were asked about their hypothetical views of TFGT (Group H, n = 12) participated in semistructured interviews. Twenty-six interviews were transcribed verbatim and analysed. Table 3 shows women's sociodemographic, medical, and family history characteristics. A chi-square test for independence (with Yates continuity correction)

indicated that no differences were noted between the groups in parity ($\chi^2 [1, n = 26] = 0.58, p = 0.45, \phi = -0.23$) or in the presence or absence of daughters ($\chi^2 [1, n = 26] = 0.67, p = 0.41, \phi = -0.24$). Two participants reported a previous cancer diagnosis, and only two were known to carry a *BRCA* mutation. Participants described in this article will be denoted by their group (A or H); C will denote participants with children and NC will denote participants with no children.

When should treatment-focused genetic testing be presented and by whom? Overall, many participants (n = 16) felt TFGT should be offered before decisions on cancer management, including surgery, were made to get "everything over in one go" (A, NC). The majority of participants (n = 14) preferred TFGT to be discussed close to diagnosis and in the context of treatment decisions being discussed.

I think not in the initial diagnosis, because that's such a shock in itself. The best time for me was when it came to discussing my treatment . . . but when it was talking about the whole theme of what my treatment would look like, in amongst that (A, C).

A number of women (n = 8), however, preferred TFGT to be presented at the time of diagnosis, despite acknowledging that the time was fraught and emotionally overwhelming. As expressed by one woman,

I think it has to come at the start as part of being diagnosed and then everything explained. When you're diagnosed, you are learning all this new language so I suppose this then has to come into play as well. Because you're asking what caused it—how did I get it? (A, NC).

No clear majority preference emerged for which healthcare professional should introduce TFGT. In particular, eight women expressed a preference for the surgeon as the best person to initiate a discussion about TFGT, six participants preferred to have the discussion with their oncologist, and six with a genetics practitioner (clinical geneticist or genetic counselor). Five preferred to involve the breast care nurse, as "someone who's consistent through your care" (A, C), and one woman did not specify a preference for a particular type of healthcare professional. Of the 14 women who preferred to have TFGT raised by their oncologist or surgeon, eight

Table 1. Satisfaction With Educational Materials

| Description | Response | |
|---|----------|----|
| | Yes | No |
| Any parts of the pamphlet that you thought should have been explained in more detail? | 10 | 7 |
| Any information not covered in the pamphlet that you think should be included? | 7 | 10 |
| Could any parts of the pamphlet be left out? | 4 | 13 |
| In your opinion, was there anything in the pamphlet that was confusing? | 7 | 10 |
| Was the tone of the pamphlet positive enough? | 15 | 2 |

| Description | Far Too Much or Too Much | About Right | Too Little or Far Too Little |
|---|--------------------------|-------------|------------------------------|
| How would you describe the amount of information in the pamphlet? | 1 | 15 | 1 |

| Description | Very | Somewhat | Not Very or Not at All |
|--|------|----------|------------------------|
| Was the pamphlet clearly laid out? | 14 | 2 | 1 |
| Was the pamphlet written in language that is easy to understand? | 14 | 2 | 1 |
| Was the pamphlet useful? | 14 | 3 | — |
| Was the pamphlet appealing to look at? | 9 | 5 | 3 |

| Description | Very Satisfied | Satisfied | Dissatisfied or Very Dissatisfied |
|--|----------------|-----------|-----------------------------------|
| How satisfied were you with the information in the pamphlet? | 10 | 6 | 1 |

N = 17

Table 2. Mean Ratings of Perceived Improvement in Understanding of TFGT

| To what extent do you think the pamphlet would have improved your understanding of . . . | Score | |
|--|-----------|-----|
| | \bar{X} | SD |
| What TFGT is | 4.3 | 0.9 |
| The purpose of TFGT | 4.5 | 0.7 |
| The relevance of TFGT in your situation | 4 | 0.9 |
| How TFGT is done | 4 | 0.9 |
| The benefits of TFGT in your situation | 4.2 | 0.9 |
| The disadvantages of TFGT in your situation | 2.8 | 1.4 |
| What it would mean if a faulty breast cancer gene was not found in me | 3.8 | 0.8 |
| What it would mean if a faulty breast cancer gene was found in me | 4.2 | 0.8 |
| What could be done if you were found to have inherited a faulty breast cancer gene | 4.3 | 0.7 |

N = 17

TFGT—treatment-focused genetic testing

Note. Higher mean ratings indicate greater improvement. Scores were 1 (not at all), 2 (a little), 3 (somewhat), 4 (quite a bit), or 5 (a lot).

wanted to be able to discuss the test result with a genetic healthcare professional when it became available, and two wanted to discuss the implications with the breast care nurse.

Because you're working in a very small time frame, it's going to be someone like the surgeon who brings it up, but if you have a way of having either the breast care nurse or genetic counselor on hand so that . . . if you've got more questions, these people can answer your questions, sit down, talk with you more, and help with decision making (H, NC).

How should treatment-focused genetic testing be presented? The vast majority of women (n = 22) preferred to receive information about the availability of TFGT during a face-to-face consultation. That enabled them to ask questions spontaneously, as one woman expressed.

I found it valuable that I had the opportunity to ask questions. It was a fairly emotional time because I'd just been diagnosed, but then having some literature to read to follow-up afterwards was also valuable (A, C).

A face-to-face consultation also provided the opportunity for a more personalized approach, including the option of having a support person present. It also allowed patients to determine the level of information they felt they could assimilate.

It's funny because I was given all the DVDs and the Web site, and I have never gone to any one of them. Even though what I do is research, so usually I would do exactly that, I was actually afraid of all

those things because I didn't want to find information that I didn't want to know. So by asking questions I had at least a feeling of control over it (A, C).

Additional written supporting information to accompany an offer of TFGT was seen as essential by most participants (n = 16), allowing them to take information home, absorb it, and formulate questions at their own pace.

I found the whole process was overwhelming and it was useful to take things away, get home, have a cup of tea, read over it, read over it again, and have it in writing in front of me (A, C).

Nine women felt a Web site would be a useful supplementary tool, and eight felt they would have benefited from watching a DVD or other audiovisual material.

What information should be presented? Although all participants agreed that they would like all the implications of TFGT relating to themselves covered, several participants expressed feeling overwhelmed by information around the time of their diagnosis, and the majority (n = 21) preferred information to be brief and to the point. Eleven women preferred a leaflet, and another 11 felt a decision aid could be helpful; however, only five participants reported that they would use a personal worksheet to help them arrive at a decision on whether or not to have TFGT.

The example information sheet provided to participants was found to provide an acceptable level of information for 15 women. Those who did not find it acceptable (n = 5) expressed a desire to discuss the information face-to-face with a healthcare professional instead of receiving educational materials. The remaining four participants did not express a viewpoint on the example provided, and six other women did not provide a response for the question.

Several participants listed specific topic areas they would like covered in the educational materials, including the purpose of TFGT (H, C); the chance of carrying a gene mutation (H, NC; A, C); other factors that may elevate a woman's breast cancer risk above that of population risk (A, C); the impact of the test result (A, C), particularly on treatment (H, NC); what the blood test involves (H, C); how long it takes for the results to be returned (A, C); that family members would need to make up their own minds about having predictive genetic testing (H, NC); and that family members may have unexpected emotional reactions to the news (A, C), such as anger and hostility.

Implications for themselves: Some women (n = 9) emphasized their desire for the information provided to focus on the impact of TFGT on themselves, the meaning of the test result, and, in particular, the impact of TFGT on their treatment options. Many participants (n = 16) expressed a desire for all available information

up front, including information on the chances of developing a second breast cancer (n = 6) and/or ovarian cancer (n = 13).

For me, I wanted to know everything. I did not know the increased risk of ovarian cancer associated with *BRCA1* and 2. That was very valuable to know that because, again, it would have made me make decisions about prophylactic oophorectomy (A, C).

Some concern arose, however, that listing the increased risks of other cancers may further frighten women at an already vulnerable time and, in some instances, may dissuade them from participating in TFGT. As a result, some participants believed that the positive outcomes listed in the description of such a test should be more clearly expressed.

Although the majority of participants (n = 16) preferred all clinically relevant information to be presented briefly, some (n = 7) preferred detailed information on surgical options to be discussed up front. Conversely, a few (n = 3) preferred the details about surgical options to be discussed only if a positive test result was received.

It's unnecessary worry to think, you know, if you read all that and you think, "Oh my God is that what I'm going to have to go through?" (A, C).

Implications for the family: Many women (n = 18) felt that the brochure should "remain focused on the woman" (A, NC), and family implications should only be mentioned briefly, with more detail provided in face-to-face genetic counseling should they receive a positive genetic test result.

I think it's probably worth mentioning in there. If you have this gene change, other members of your family may also have inherited this. You don't need to go into full-scale statistics (H, NC).

Stage 2: Development and Pilot Evaluation of the Pamphlet

Letters of invitation were sent to 23 of the 26 women who participated in stage 1. Three participants from stage 1 were not approached (two women declined approach for pilot testing at stage 1 and another woman was diagnosed with metastatic breast cancer after her stage 1 interview). Eighteen women opted into stage 2 (response rate of 78%), with one woman unable to be scheduled for

interview. Seventeen women, 10 from Group A and 7 from Group H, participated in the stage 2 interviews.

Statistical analysis (two-sided Fisher's exact test) confirmed that no differences were noted between Group A and Group H regarding satisfaction with the information in the pamphlet (p = 0.412) or in the emotional impact of the pamphlet ("worry or concern," p = 0.25; "upset or sad," p = 0.998). An independent samples t test was conducted to compare the scores for perceived improvement in understanding for Group A and Group H. No significant differences were noted in scores for Group A (\bar{X} = 36.3, SD = 3.77) and Group H (\bar{X} = 35.57, SD = 6.11), $t(15) = 0.305$, $p = 0.764$ (two-tailed; 95% confidence interval [-4.36, 5.82]). Therefore, the findings for all stage 2 outcomes are reported for the entire sample rather than for each group separately.

Table 3. Participant Demographics by Group

| Variable | Group A (N = 14) | | Group H (N = 12) | | Total Sample (N = 26) | |
|---|---------------------|----|---------------------|----|--------------------------|----|
| | \bar{X} | SD | \bar{X} | SD | \bar{X} | SD |
| Age at diagnosis (years) | 40 | 5 | 42 | 5 | 41 | 5 |
| Age at interview (years) | 42 | 5 | 43 | 5 | 42 | 5 |
| Variable | n | n | n | | | |
| Highest level of education | | | | | | |
| High school | 4 | 3 | 7 | | | |
| Post-high school qualification | 10 | 9 | 19 | | | |
| Marital status | | | | | | |
| Married or cohabiting | 11 | 9 | 20 | | | |
| Not married | 3 | 3 | 6 | | | |
| Biological children | | | | | | |
| Yes | 9 | 5 | 14 | | | |
| No | 5 | 7 | 12 | | | |
| Daughter(s) | | | | | | |
| Yes | 8 | 4 | 12 | | | |
| No | 6 | 8 | 14 | | | |
| Previous cancer ^a | 2 | – | 2 | | | |
| Family history of breast and/or ovarian cancer | | | | | | |
| Yes | 14 | 5 | 19 | | | |
| No | – | 7 | 7 | | | |
| Mutation status | | | | | | |
| BRCA carrier | 2 | – | 2 | | | |
| Inconclusive result ^b | 12 | – | 12 | | | |

^a Hodgkin lymphoma; melanoma

^b If a deleterious gene mutation is not detected in *BRCA1* or *BRCA2*, and the participant does not have a family history of breast and/or ovarian cancer, her breast cancer unlikely is caused by an inherited mutation in a breast cancer protection gene. However, as not all breast cancer protection genes have been discovered, if a participant does have a strong family history of breast and/or ovarian cancer, she may carry a mutation in an as yet undiscovered predisposition gene. For that reason, the result is termed *inconclusive* and the participant and her family still may be at increased risk of breast and ovarian cancer.

TFGT—treatment-focused genetic testing

Note. Group A denotes women who had TFGT (actual decision making). Group H denotes women who did not have TFGT (hypothetical decision making).

Satisfaction with the pamphlet: Almost all women (n = 16) reported being satisfied or very satisfied with the information provided, or reported that the amount of information in the pamphlet was about right (n = 15). Most women (n = 14) reported that the pamphlet would have been very useful around the time of diagnosis, with three stating that it would have been somewhat useful.

Participants mentioned one or more parts of the pamphlet where more detail was needed: the implications of TFGT (n = 6), surgical options (n = 3), and more specific information about the timing of TFGT (n = 3). Women reported that what they liked best about the pamphlet was the format, including the question-and-answer style (n = 3), the layout and order of topics (n = 2), and the highlighting of important points (n = 1). Aspects of the pamphlet that women liked least also related to presentation, and included the cover photograph (n = 4), the “amateurish” format (n = 1), and the dated photograph on an inside page (n = 3). The section of the pamphlet that was identified most frequently as confusing was “what if a gene fault is not found or is found in me?” (n = 3). Suggestions for improving the pamphlet included clarifying the timing of genetic testing (n = 2), providing a contact number or the location of a genetic counselor (n = 2), clarifying the section “not finding a faulty gene” (n = 1), removing the TFGT acronym (n = 1), and modernizing the layout (n = 1).

Perceived improvement in understanding: Most participants (n = 15) reported that the booklet had improved their understanding. The largest perceived improvement in understanding was in relation to the purpose of TFGT. The smallest perceived improvement in understanding was in relation to the disadvantages of TFGT in the woman’s situation.

Emotional impact of the pamphlet: Most participants (n = 13) reported feeling not at all worried or concerned by reading the pamphlet, whereas some (n = 4) reported feeling a little or somewhat worried or concerned. Of the four who were worried after reading the pamphlet, three reported in the open-ended question that it reminded them about the time of their breast cancer diagnosis (n = 4) and one woman raised concerns about her relatives (n = 1). Almost all participants (n = 15) reported that reading the pamphlet did not make them feel at all sad or upset.

Perceived importance of information about treatment-focused genetic testing: Fifteen participants reported that TFGT was very important for women in their situation. Eleven women perceived that their clinicians believed that TFGT was very important, and five women said that it was somewhat or not at all important. Seven participants reported that the pamphlet would have been enough to make a decision about TFGT soon after diagnosis. Some participants who reported that the

pamphlet would not have been enough said that they would, in addition, have needed to speak to someone about TFGT (n = 10), with six indicating they would want to speak to a healthcare professional or search online for more information, and four not indicating a preference.

Discussion

All women who participated in the study agreed that TFGT should be offered before final decisions on cancer treatment options are made. Fifty-four percent of the women preferred TFGT to be discussed after diagnosis at a time when treatment decisions are being discussed; another 31% preferred TFGT to be presented at the time of diagnosis. The findings contrast with qualitative data from focus groups conducted with 13 women who were younger than age 40 when diagnosed with breast cancer, ascertained through a familial cancer clinic, and subsequently identified as *BRCA1* or *BRCA2* mutation carriers (Ardern-Jones et al., 2005). Although a wide range of views regarding the preferred timing of an offer of TFGT was identified, the majority of women in Ardern-Jones et al. (2005) expressed the view that an offer of genetic testing around the time of diagnosis might add too much stress at an already stressful time.

Several factors may account for the differences in findings. First, the women in the Ardern-Jones et al. (2005) study had undergone genetic testing two months to 10 years after their diagnosis and, therefore, most had completed their cancer treatment at the time of genetic testing. Consequently, all of the women were providing their views on hypothetical TFGT. By contrast, more than 54% of the women in the current study had actually undergone TFGT to inform their surgical and/or radiotherapy decisions. Women who are reflecting on their actual rather than hypothetical experience of TFGT may perceive tangible benefits from the process, and that may make it more likely for them to express a preference for TFGT at diagnosis or shortly after diagnosis when treatment options are being discussed. Second, the women in the Ardern-Jones et al. (2005) study were interviewed one to seven years following their diagnosis, and women’s attitudes to TFGT may change over time; in contrast, in the current study, 46% had been diagnosed within the previous year and were therefore closer to the time of decision making. Finally, all women in the Ardern-Jones et al. (2005) study had qualified for genetic testing and were likely to have a family history suggestive of the presence of a hereditary breast and/or ovarian cancer. The current study, however, included a substantial proportion of women without a strong family history, which may make them less likely to anticipate being a carrier of a *BRCA* mutation and the associated emotional impact, which in turn

may account for their preference to have TFGT at the time of diagnosis.

Although additional written information was seen as essential by most participants, the vast majority of women preferred to receive information about TFGT during a face-to-face consultation with a healthcare professional. Those findings concur with Vadaparampil et al. (2009), who concluded that a referral letter alone from the woman's surgeon may not be the most effective means of informing patients about TFGT. The current study's finding is not surprising, however, given that research into patients' preferred communication strategies consistently show that patients prefer to receive health-related information as part of an individual consultation with an expert (Andrews et al., 2006; Meiser, Mitchell, McGirr, Van Herten, & Schofield, 2005; Thewes et al., 2005). However, very little is currently known about whether an individual consultation with an expert is always needed to achieve informed patient choices. For example, a previous randomized, controlled trial involving women at low risk of being *BRCA* mutation carriers found that a computer program was more effective in improving knowledge of breast cancer and genetic testing, facilitating more accurate risk perceptions, and lowering anxiety compared to standard counseling (Green et al., 2004). Given the likely increasing burden on familial cancer services and rising costs of health care, future prospective studies are required to determine the most effective ways of offering information about TFGT.

No clear majority preference was established for which type of healthcare professional should provide information on TFGT. Two previous studies have produced divergent results, with patients interviewed in the Arden-Jones et al. (2005) study reporting a preference for receiving information about genetic testing from a genetics practitioner. Those patients all had attended a familial cancer clinic where they saw a genetics practitioner. In a prospective study, the majority of patients with breast cancer who were offered genetic testing at the start of their radiotherapy preferred their surgeon to present the genetic information (Schlich-Bakker, ten Kroode, Warlam-Rodenhuis, van den Bout, & Ausems, 2007). In the current study, the majority also preferred all clinically relevant information to be presented briefly, with some preferring details on surgical options to be discussed up front. Taken together, the findings suggest that the type of healthcare professional is not critical as long as he or she is in a position to present the clinically relevant information to the woman, answer her questions, and gauge the level of detail she feels she can assimilate.

The study also provided detailed information about the preferred content and level of detail of any education materials women wished to receive. In particular, the majority of women preferred the information to be

brief. Women emphasized their need for information on the impact of TFGT for themselves—particularly in regard to their treatment options—whereas family implications should be addressed later in face-to-face genetic counseling should the woman prove to be a gene mutation carrier. The general perception was that the pilot-tested educational material would have been useful when making choices about TFGT and would have improved women's understanding of TFGT. The majority did not think the pamphlet had a negative emotional impact and it was generally well received.

Although the pilot study has provided timely preliminary evidence on the value of a brief written resource in preparing women for decision making about TFGT, inferring whether the educational materials alone would be as effective, as standard pretest genetic counseling is unknown. Future prospective studies among larger samples are required to determine the most effective ways of offering information about genetic testing to patients around the time of their breast cancer diagnosis. Suggested improvements to the pamphlet were incorporated after pilot testing, including modernizing the presentation and photographs, removing the TFGT acronym, and clarifying confusing content. The new resource currently is being used in a randomized, controlled trial assessing the efficacy of brief educational materials (intervention) compared to standard pretest genetic counseling (control) among women newly diagnosed with breast cancer considering genetic testing. A variety of psychological, behavioral, and decision-related outcomes are being assessed.

Limitations

The study contained limitations that should be noted. First, this was a retrospective study in which women were asked to reflect on their actual or hypothetical attitudes toward TFGT. Second, the current sample of women was highly motivated and well educated. Third, the sample size was relatively small, although acceptable for a qualitative inquiry. On the other hand, a key strength of the study was that the views of women with and without a strong family history of breast or ovarian cancer were included. The current sample is likely to represent more fully the range of patients at elevated risk of carrying a *BRCA* mutation who will be targeted by TFGT in the near future, as other high-risk features (e.g., breast cancer pathology) in addition to family history are incorporated into genetic testing criteria.

Conclusions

Optimal management of young women with breast cancer with high-risk features will require that genetic risk information be available up front at diagnosis and prior to treatment so that it may be used to inform

surgical and other treatment decisions. Therefore, the development of an educational resource about TFGT is very timely. This qualitative inquiry and pilot study has provided preliminary evidence for the efficacy of a brief written resource designed to educate women newly diagnosed with breast cancer about TFGT.

Implications for Nursing

Healthcare providers, including oncology nurses, surgeons, and oncologists, need to be appropriately equipped to educate women about genetic testing when those patients already are grappling with the emotional impact of a breast cancer diagnosis. This study represents a step toward that goal as it has produced a brief educational resource that will assist oncology nurses and other healthcare professionals to provide appropriate decisional support to women newly diagnosed with breast cancer and considering genetic testing.

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